Newborn Screening Program Disorders

Effective January 1, 2006, Utah newborns are screened for the following disorders:

METABOLIC DISORDERS:

- **Biotinidase deficiency:** a recessive disorder of biotin metabolism.
- **Galactosemia:** a recessively inherited genetic disorder in which the individual is completely or partially incapable of normal metabolism of galactose due to a deficiency of the galactose-1-phosphate uridyltransferase enzyme.

Organic Acid Disorders: recessive disorders resulting from an enzyme deficiency in the intermediary metabolism of amino acids or fatty acids.

- Beta-Ketothiolase Deficiency
- Glutaric Acidemia, Type 1
- Isobutyryl CoA Dehydrogenase Deficiency
- Isovaleric Acidemia
- Malonic Aciduria
- Maple Syrup Urine Disease
- Methylmalonic Acidemias
- Propionic Acidemia
- 3-Hydroxy-3-Methylglutaryl (HMG) CoA Lyase Deficiency
- 2-Methyl-3-Hydroxybutyryl CoA Dehydrogenase Deficiency
- 2-Methylbutyryl CoA Dehydrogenase Deficiency
- Multiple Carboxylase Deficiency

Amino acid disorders: recessive disorders resulting from an enzyme deficiency needed for amino acid metabolism or transport.

- Arginase Deficiency
- Argininosuccinate Lyase Deficiency (ASA)
- Citrullinemia
- Homocystinuria
- Hyperphenylalanemia, including Phenylketonuria (PKU)
- Tyrosinemia

Fatty Acid Oxidation Disorders: recessive disorders resulting from an enzyme deficiency needed for the break down of fatty acids.

- Carnitine uptake/transport defects
- Multiple Acyl-CoA Dehydrogenase Deficiency (MADD)
- Short Chain Acyl-CoA Dehydrogenase Deficiency (SCAD)
- Medium Chain Acyl-CoA Dehydrogenase Deficiency (MCAD)
- Long chain 3 Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
- Very long Chain Acyl-CoA Dehydrogenase Deficiency (VLCAD)
- Carnitine-Acylcarnitine Translocase Deficiency
- Carnitine Palmitoyl Transferase-1 Deficiency

ENDOCRINE DISORDERS:

- **Congenital Adrenal Hyperplasia (CAH):** a genetic disorder in which there are defects in the enzymes of the adrenal cortex required for the biosynthesis of adrenal corticosteroids.
- **Congenital Hypothyroidism:** a disorder in which the newborn is unable to secrete or produce thyroxine normally.

HEMOGLOBIN DISORDERS:

Sickle cell disease and other hemoglobinopathies: recessively inherited genetic defects of the structure of hemoglobin found in red blood cells.

- FABarts (Alpha Thalassemia carrier)
- FAS (Sickle Cell carrier)
- FAC, D, or E (Carrier trait)
- FS, FC, FE, FSC (Actual disease state)

These are disorders that may have significant mortality and morbidity when not diagnosed pre-symptomatically and may not be consistently identified clinically in the neonatal period. Early detection and treatment may improve the health and development of newborns identified with these disorders.

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